Proper Genetic Counseling Prior to Amniocentesis

To the Editor:

I read with great interest the article by Chen et al [1]. They concluded that, in view of the effective use of amniocentesis as a diagnostic procedure for Down syndrome, efforts should be made to use more efficient prenatal screening programs and to reduce the number of unnecessary amniocentesis procedures. So-called elective genetic amniocentesis should also be classified under "unnecessary amniocentesis". Although amniocentesis no longer results in a significant improvement in the detection rate of Down syndrome in the Taiwanese population, I do not agree that more powerful screening programs such as first-trimester maternal serum screening in conjunction with nuchal translucency and nasal bone measurement by early ultrasound examination are "required". If that is the consensus of the Taiwan Amniocentesis Collaborative Study Group, then all practitioners in Taiwan should follow that recommendation, which has not been tested and verified. One should also bear in mind that many newly developed prenatal screening programs are still considered experimental. Perhaps a more appropriate statement would be that these more powerful screening programs should be "encouraged".

Prenatal screening programs for Down syndrome, for example, in England and Wales, that were based on maternal serum biochemistry or ultrasound were more effective and efficient than screening programs that used advanced maternal age alone [2]. This is well known. Genetic ultrasound for detection of trisomy 21 was considered a safe and cost-effective procedure in the US [3]. In the Israeli population, new genetic methods to assess Down syndrome risk did not improve the rate of detection or reduce the number of amniocenteses, but was accompanied by an increased cost per case detected [4]. Many women choose noninvasive prenatal testing before making a decision regarding whether or not to undergo amniocentesis [5]. Therefore, in making prenatal testing guidelines, women should be allowed to make informed decisions regarding the use of invasive testing that are reflective of their own values and preferences [6]. Patient anxiety (and doctor anxiety) should not be used as an excuse to perform amniocentesis. It is shameful for anyone to carry out elective genetic amniocentesis without a specific indication. Proper genetic counseling is important and should be emphasized.

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